ZNF341 gene

zinc finger protein 341

Normal Function

The *ZNF341* gene provides instructions for making a transcription factor, which is a protein that attaches (binds) to specific regions of DNA and helps control the activity of particular genes. The ZNF341 protein is thought to regulate the activity of the *STAT1* and *STAT3* genes, controlling production of the STAT1 and STAT3 proteins, respectively. Both proteins are involved in the immune system. They control pathways in cells that help fight foreign invaders such as viruses, bacteria, and fungi. The STAT3 protein, in particular, transmits signals for the maturation of immune system cells, especially T cells and B cells. STAT3 is also involved in normal development and maintenance of bones and other tissues.

Researchers suspect that the ZNF341 protein controls the activity of other genes, although they have not been identified.

Health Conditions Related to Genetic Changes

Autosomal dominant hyper-IgE syndrome

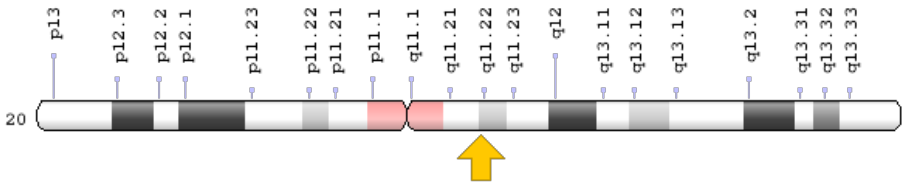
At least five mutations in the *ZNF341* gene have been found to cause a condition similar to autosomal dominant hyper-IgE syndrome (AD-HIES), which is a disorder of the immune system characterized by recurrent skin and lung infections and abnormally high levels of an immune system protein called immunoglobulin E (IgE) in the blood. AD-HIES is usually caused by *STAT3* gene mutations and follows an autosomal dominant pattern of inheritance, which means one altered copy of the gene is sufficient to cause the disorder. In contrast, the condition caused by *ZNF341* gene mutations follows an autosomal recessive pattern of inheritance, which means both copies of the gene must be altered for immune system problems to develop.

The *ZNF341* gene mutations that cause an AD-HIES-like condition result in production of an abnormally short ZNF341 protein or production of no protein. With little or no ZNF341 protein, production of the STAT1 and STAT3 proteins is impaired. A shortage of functional STAT3 blocks the maturation of T cells (specifically a subset known as Th17 cells) and other immune cells. The resulting immune system abnormalities make people with AD-HIES highly susceptible to infections, particularly bacterial and fungal infections of the lungs and skin. A shortage of STAT1 protein is not thought to contribute to immune system problems in affected individuals.

Chromosomal Location

Cytogenetic Location: 20q11.22, which is the long (q) arm of chromosome 20 at position 11.22

Molecular Location: base pairs 33,731,657 to 33,792,269 on chromosome 20 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ZNF341 gene

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ZNF341%5BTIAB%5D%29+OR+%28zinc+finger+protein+341%5BTIAB%5D%29%29+OR+%28ZNF341+gene%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- ZINC FINGER PROTEIN 341
  http://omim.org/entry/618269

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ZNF341%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:15992
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:84905

- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/84905
- UniProt
  https://www.uniprot.org/uniprot/Q9BYN7

Sources for This Summary

- Béziat V, Li J, Lin JX, Ma CS, Li P, Bousfiha A, Pellier I, Zoghi S, Baris S, Keles S, Gray P, Du N, Wang Y, Zerbib Y, Lévy R, Leclercq T, About F, Lim AI, Rao G, Payne K, Pelham SJ, Avery DT, Deenick EK, Pillay B, Chou J, Guery R, Belkadi A, Guérin A, Migaud M, Rattina V, Ailal F, Benhsaien I, Bouaziz M, Habib T, Chaussabel D, Marr N, El-Benna J, Grimbacher B, Wargon O, Bustamante J, Boisson B, Müller-Fleckenstein I, Fleckenstein B, Chandesris MO, Titeux M, Fraitag S, Alyanakian MA, Leruez-Ville M, Picard C, Meyts I, Di Santo JP, Hovnanian A, Somer A, Ozen A, Rezaei N, Chatila TA, Abel L, Leonard WJ, Tangye SG, Puel A, Casanova JL. A recessive form of hyper-IgE syndrome by disruption of ZNF341-dependent STAT3 transcription and activity. Sci Immunol. 2018 Jun 15;3(24). pii: eaat4956. doi: 10.1126/sciimmunol.aat4956.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/29907691
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6141026/
- Frey-Jakobs S, Hartberger JM, Fliegauf M, Bossen C, Wehmeyer ML, Neubauer JC, Bulashevska A, Proietti M, Fröbel P, Nöltner C, Yang L, Rojas-Restrepo J, Langer N, Winzer S, Engelhardt KR, Glocker C, Pfeifer D, Klein A, Schäffer AA, Lagovsky I, Lachover-Roth I, Béziat V, Puel A, Casanova JL, Fleckenstein B, Weidinger S, Kilic SS, Garty BZ, Etzioni A, Grimbacher B. ZNF341 controls STAT3 expression and thereby immunocompetence. Sci Immunol. 2018 Jun 15;3(24). pii: eaat4941. doi: 10.1126/sciimmunol.aat4941.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/29907690
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6173313/
- OMIM: ZINC FINGER PROTEIN 341
  http://omim.org/entry/618269
- Zhang Q, Boisson B, Béziat V, Puel A, Casanova JL. Human hyper-IgE syndrome: singular or plural? Mamm Genome. 2018 Aug;29(7-8):603-617. doi: 10.1007/s00335-018-9767-2. Epub 2018 Aug 9. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/30094507
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6317873/

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/ZNF341

Reviewed: August 2019
Published: June 23, 2020

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services